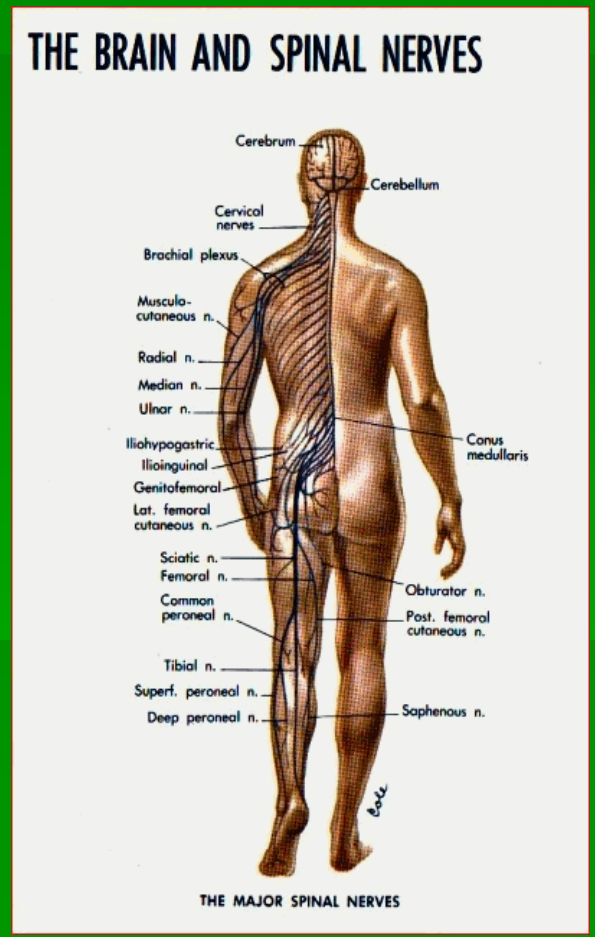
Spinal Muscular Atrophy

What is it?

Spinal Muscular Atrophy (SMA)

- Genetic, motor neuron disease caused by progressive degeneration of motor neurons in the spinal cord
- Manifestation of weakness due to loss of the motor neurons of the spinal cord
- Caused by mutation of the SMN gene
- Term applied to a number of different disorders
- Wide range of severity affecting infants through adults



What Causes SMA?

- SMA is an autosomal recessive genetic disease
  - In order to be affected by SMA, both parents must be carriers of the gene and both must pass this gene on to the child
- An individual with SMA has a missing or mutated gene *(SMN1, or survival motor neuron 1)* which affects motor neurons and how they function
  - Without the protein nerve cells atrophy, shrink and eventually die, resulting in muscle weakness

Who is Affected?

- SMA is one of the most prevalent genetic disorders
- SMA can strike anyone of any age, race or gender
- One in every 6,000 babies is born with SMA
- Children diagnosed before age two, 50 percent will die before their second birthday
- One in every 40 people carries the gene that causes SMA
  - The child of two carriers has a one in four chance of developing SMA

Types of SMA

- **Type 1 Infantile SMA - or Werdnig-Hoffmann disease**
  - generally 0-6 months
  - most severe
  - inability to ever maintain an independent sitting position
- **Type 2 Intermediate SMA**
  - generally 7-18 months
  - children who are never able to stand and walk, but are able to maintain a sitting position at least some time in their life
- **Type 3 Juvenile SMA - or Kugelberg-Welander disease**
  - generally >18 months
  - Individuals are able to walk at some time in life
- **Type 4 Adult SMA**
  - Weakness usually begins in late adolescence in tongue, hands, or feet then progresses to other areas of the body
  - Course of disease is much slower and has little or no impact on life expectancy

Symptoms

- **Type 1 Infantile SMA**, *Werdnig-Hoffmann disease (0-6mths)*
  - floppiness of the limbs and trunk
  - feeble movements of the arms and legs
  - swallowing and feeding difficulties
  - impaired breathing
- **Type 2 Intermediate** *(7-18mths)*
  - respiratory problems
  - floppy limbs
  - decreased or absent deep tendon reflexes
  - twitching of arm, leg, or tongue muscles

Symptom Cont.

- **Type 3 Juvenile SMA**, *Kugelberg-Welander disease (> 18mths)*
  - abnormal manner of walking
  - difficulty running
  - climbing steps or rising from a chair
  - slight tremor of the fingers
- **Type 4 Adult SMA**
  - weakness of muscles in the tongue and face
  - difficulty swallowing
  - speech impairment
  - excessive development of the mammary glands in males

How is it diagnosed?

- If there is evidence of degeneration in lower motor neurons in the spinal cord and brainstem
- A history of motor difficulties
- Evidence of motor unit disease on physical examination
- SMN gene test
  - determines if there is a copy of the SMN1 gene by looking for its sequences
- Electromyography (EMG) or muscle biopsy

Prognoses

- Varies depending on the type of SMA and the degree of respiratory function
- The patient's condition tends to deteriorate over time
- Life expectancy depends on the type you have and how it affects your breathing.
- There is no cure.

Treatment

- All treatment is symptomatic and specific to the type of SMA
- Some specific treatments may include:
  - physical therapy
  - medicines
  - treating respiratory infections
  - treating pneumonia
  - treating curvature of the spine
  - orthotic supports
  - genetic counseling

Organizations for Support

FightSMA/Andrew's Buddies
1807 Libbie Avenue
Suite 104
Richmond, VA 23226
heatherlennon@fightsma.com
http://www.fightsma.org
Tel: 804-515-0080
Fax: 804-515-0081

Families of Spinal Muscular Atrophy
P.O. Box 196
Libertyville, IL 60048-0196
sma@fsma.org
http://www.curesma.org
Tel: 847-367-7620 800-886-1762
Fax: 847-367-7623

Spinal Muscular Atrophy Foundation
119 West 72nd St.
#187
New York, NY 10023
info@smafoundation.org
http://www.smafoundation.org
Tel: 877-FUND-SMA (877-386-3762)
646-253-7100
Fax: 212-247-3079

March of Dimes Birth Defects Foundation
1275 Mamaroneck Avenue
White Plains, NY 10605
askus@marchofdimes.com
http://www.marchofdimes.com
Tel: 914-428-7100 888-MODIMES (663-4637)
Fax: 914-428-8203

Muscular Dystrophy Association
3300 East Sunrise Drive
Tucson, AZ 85718-3208
mda@mdausa.org
http://www.mda.org
Tel: 520-529-2000 800-344-4863
Fax: 520-529-5300

Kennedy's Disease Association
P.O. Box 1105
Coarsegold, CA 93614-1105
info@kennedysdisease.org
http://www.kennedysdisease.org
Tel: 559-658-5950

Sources

- http://en.wikipedia.org/wiki/Spinal_muscular_atrophy
- http://www.ninds.nih.gov/disorders/sma/sma.htm
- http://www.ninds.nih.gov/disorders/sma/sma.htm#Is_there_any_treatment
- http://www.fsma.org/sma_facts.shtml
- http://www.fsma.org/booklet.shtml#causes